STATEMENT OF SCOPE
WISCONSIN DEPARTMENT OF HEALTH SERVICES

CHAPTER: DHS 115

RELATING TO: SCREENING NEWBORNS FOR CONGENITAL AND METABOLIC DISORDERS

RULE TYPE: EMERGENCY AND PERMANENT

SCOPE TYPE: ORIGINAL

FINDINGS OF EMERGENCY:

The department of health services ("the department") finds that an emergency exists and that the adoption of an emergency rule may be necessary for the immediate preservation of the public health, safety, and welfare. The facts constituting the emergency are as follows:

1. Section 253.13 (1) Stats., requires attending physicians and nurses licensed under s 444.15, Stat., to cause every infant born in each hospital or maternity home, prior to the infant's discharge to be subjected to tests for congenital and metabolic disorders, as specified in rules promulgated by the department. If the infant is born elsewhere than in a hospital or maternity home, the attending physician, nurse licensed under s. 441.15. Stats., or birth attendant who attended the birth shall cause the infant, within one week of birth, to be subjected to these tests.
2. Section DHS 115.04 lists the disorders for which newborns must be tested under s. 253.13 (1), Stats.
3. The immediate addition of Pompe disease is important to the quality of life, well-being, and public health of Wisconsin residents. The earliest diagnosis and initiation of treatment of Pompe can make a difference between an infant's survival and positive outcomes and death and severe disability. Infantile-onset Pompe disease presents with failure to thrive, muscle weakness, hypotonia, respiratory distress and cardiomyopathy at a median age of 4 months. Untreated, the condition is generally fatal by 2 years of age. Initiation of treatment prior to symptom onset improves survival and outcomes. Late-onset Pompe disease, which is also diagnosed by the newborn screening test, can be treated to slow or prevent development of progression of disease.
4. Preservation of the health, safety, and welfare of newborns and infants thereby necessitates promulgating an emergency rule prior to the time it would take effect if the department complied with all of the procedures under Ch. 227, Stats.

SUMMARY

1. Description of rule objective/s

The objective of the rule is to add Pompe disease to the panel of congenital and metabolic disorders for which newborns shall be tested in accordance with s. 253.13 (1). The addition of Pompe disease to the newborn screening panel was based on the consideration and recommendations of the Wisconsin Newborn Screening Program ("NBS") and the Secretary's Advisory Committee on Newborn Screening ("SACNBS"). Based upon those recommendations, Secretary-designee Palm approved adding Pompe disease to the newborn screening panel.

2. Existing policies relevant to the rule

Section 253.13 (1), Stats., requires every infant born in a hospital or maternity home or other place to be screened for congenital and metabolic disorders as specified in rule by the department. In order to fulfill its statutory duty to identify those disorders by rule, s, DHS 115.06 provides that the department seek "the advice and guidance of medical consultants, staff of the state laboratory and other persons who have expertise and

experience in dealing with congenital and metabolic disorders" to determine whether to add or delete disorders to the newborn screening panel.

The NBS Umbrella Committee and its subcommittees—which are comprised of a variety of medical practitioners, experts on genetics, pediatrics and medical ethics, and various advocacy organizations—meet regularly to review and evaluate program processes and make recommendations to the department's secretary with respect to adding or deleting a condition from the newborn screening panel. The NBS Umbrella Committee recommendations are then forwarded to the SACNBS—comprised of experts on medicine, statistics, epidemiology, medical ethics, and legal, social, and policy—which advises the secretary on policy issues related to newborn screening panel of conditions and makes recommendations on additions to the newborn screening panel.

The SACNBS recommendations are then forwarded to the department's secretary, who makes a final determination based on the NBS Umbrella Committee and SACNBS recommendations.

3. Policies proposed to be included in the rule

Pompe disease is a rare (approximately 1 in 40,000 births), inherited condition. It is considered a lysosomal storage disorder because people with Pompe have lysosomes (the recycling center of each cell) that cannot break down certain types of complex sugars. Pompe disease is caused by mutations in a gene that make an enzyme called acid alpha-1, 4-glugosidase (GAA) or acid maltase. Normally, the body uses GAA to break down glycogen, a stored form of sugar used for energy. The GAA gene is responsible for making this enzyme. Without the proper function of GAA, glycogen that enters into the lysosome is not broken down, but continues to build up and disrupts the functioning of cells. Excessive amounts of lysosomal glycogen accumulate everywhere in the body, but the cells of the heart and skeletal muscles are the most seriously affected.

On August 26, 2019 Pompe disease was nominated by a family with children who have Pompe Disease and cosponsored by another family advocate. A NBS subcommittee and the NBS Umbrella Committee considered the nomination at meetings on September 6 and December 6, 2019, and recommended adding Pompe disease to the newborn screening panel. The NBS Umbrella Committee recommendation was then forwarded to the SACNBS, which considered the nomination on March 6, 2020, and voted in support of adding Pompe disease to the newborn screening panel. On May 26, 2020, Secretary-designee Palm approved the SACNBS's recommendation to add Pompe disease to the newborn screening panel.

4. Analysis of policy alternative

Section 253.13 (1), Stats. requires that every infant born in each hospital or maternity home, prior to the infant's discharge, be tested for congenital and metabolic disorders, as specified in rules promulgated by the department. Experts on the NBS Umbrella Committee and SACNBS recommended adding Pompe disease to the newborn screening panel, and Secretary-designee Palm approved adding it to that panel and promulgating a rule to add Pompe disease to the newborn screening panel.[1] Therefore, there are no reasonable alternatives to the proposed rulemaking.

5. Statutory authority for the rule

a. Explanation of authority to promulgate the proposed rule

The department's authority to promulgate the proposed rule is provided in ss. 227.11 (2) and 253.13 (1) and (2), Stats.

b. Statute/s that authorize/s the promulgation of the proposed rule

[1] *See* Letter from Secretary-designee Palm (May 26, 2020), available at https://www.dhs.wisconsin.gov/newbornscreening/secretary-letter-pompe.pdf

Section 227.11 (2), Stats:

Rule−making authority is expressly conferred on an agency as follows:
(a) Each agency may promulgate rules interpreting the provisions of any statute enforced or administered by the agency, if the agency considers it necessary to effectuate the purpose of the statute, but a rule is not valid if the rule exceeds the bounds of correct interpretation. All of the following apply to the promulgation of a rule interpreting the provisions of a statute enforced or administered by an agency:
1. A statutory or nonstatutory provision containing a statement or declaration of legislative intent, purpose, findings, or policy does not confer rule−making authority on the agency or augment the agency's rule−making authority beyond the rule−making authority that is explicitly conferred on the agency by the legislature.
2. A statutory provision describing the agency's general powers or duties does not confer rule−making authority on the agency or augment the agency's rule−making authority beyond the rule− making authority that is explicitly conferred on the agency by the legislature.
3. A statutory provision containing a specific standard, requirement, or threshold does not confer on the agency the authority to promulgate, enforce, or administer a rule that contains a standard, requirement, or threshold that is more restrictive than the standard, requirement, or threshold contained in the statutory provision.
(b) Each agency may prescribe forms and procedures in connection with any statute enforced or administered by it, if the agency considers it necessary to effectuate the purpose of the statute, but this paragraph does not authorize the imposition of a substantive requirement in connection with a form or procedure.
(c) Each agency authorized to exercise discretion in deciding individual cases may formalize the general policies evolving from its decisions by promulgating the policies as rules which the agency shall follow until they are amended or repealed. A rule promulgated in accordance with this paragraph is valid only to the extent that the agency has discretion to base an individual decision on the policy expressed in the rule.
(d) An agency may promulgate rules implementing or interpreting a statute that it will enforce or administer after publication of the statute but prior to the statute's effective date. A rule promulgated under this paragraph may not take effect prior to the effective date of the statute that it implements or interprets.
(e) An agency may not inform a member of the public in writing that a rule is or will be in effect unless the rule has been filed under s. 227.20 or unless the member of the public requests that information.

Section 253.13 (1) and (2), Stats:

(1) TESTS; REQUIREMENTS. The attending physician or nurse licensed under s. 441.15 shall cause every infant born in each hospital or maternity home, prior to its discharge therefrom, to be subjected to tests for congenital and metabolic disorders, as specified in rules promulgated by the department. If the infant is born elsewhere than in a hospital or maternity home, the attending physician, nurse licensed under s. 441.15, or birth attendant who attended the birth shall cause the infant, within one week of birth, to be subjected to these tests.

(2) TESTS; DIAGNOSTIC, DIETARY AND FOLLOW-UP COUNSELING PROGRAM; FEES. The department shall contract with the state laboratory of hygiene to perform the tests specified under this section and to furnish materials for use in the tests. The department shall provide necessary diagnostic services, special dietary treatment as prescribed by a physician for a patient with a congenital disorder as identified by tests under sub. (1) or (1m) and follow-up counseling for the patient and his or her family. The department shall impose a fee, by rule, for tests performed under this section sufficient to pay for services provided under the contract. The department shall include as part of the fee established by rule amounts to fund the provision of diagnostic and counseling services, special dietary treatment, and periodic evaluation of infant screening programs, the costs of consulting with experts under sub. (5), the costs of administering the hearing screening program under s. 253.115, and the costs of administering the congenital disorder program under this section and shall credit these amounts to the appropriation accounts under s. 20.435 (1) (ja) and (jb).

c. Statute/s or rule/s that will affect the proposed rule or be affected by it

DHS 115.06 Criteria for adding and deleting conditions.

In determining which disorders are to be added or deleted from s. DHS 115.04, the department shall seek the advice and guidance of medical consultants, staff of the state laboratory and other persons who have expertise and experience in dealing with congenital and metabolic disorders. Criteria to be considered in adding or deleting disorders shall include all of the following:

(1) Characteristics of the specific disorder, including disease incidence, morbidity and mortality.
(2) The availability of effective therapy and potential for successful treatment.
(3) Characteristics of the test, including sensitivity, specificity, feasibility for mass screening and cost.
(4) The availability of mechanisms for determining the effectiveness of test procedures.
(5) Characteristics of the screening program, including the ability to collect and analyze specimens reliably and promptly, the ability to report test results quickly and accurately and the existence of adequate follow−up and management programs.
(6) The expected benefits to children and society in relation to the risks and costs associated with testing for the specific condition.

In addition, as a part of the nomination review process, the department sought the advice and guidance of medical consultants and experts via the NBS Umbrella Committee and SACNBS. The SACNBS recommended adding this condition to the newborn screening panel. Prior to the committee's recommendation and report submission to the Secretary of DHS, the following criteria were reviewed:

1) Mandated testing should be limited to conditions that cause serious health risks in childhood that are unlikely to be detected and prevented in the absence of newborn screening.
2) For each condition, there should be information about the incidence, morbidity and mortality, and the natural history of the disorder.
3) Conditions identified by newborn screening should be linked with interventions that have been shown in well-designed studies to be safe and effective in preventing serious health consequences.
4) The interventions should be reasonably available to affected newborns.
5) Appropriate follow-up should be available for newborns who have a false positive newborn screen.
6) The characteristics of mandated tests in the newborn population should be known, including specificity, sensitivity, and predictive value or other convincing medical evidence (experience, natural history, or literature).
7) If a new sample collection system is needed to add a disorder, reliability and timeliness of sample collection must be demonstrated.
8) Before a test is added to the panel, the details of reporting, follow-up, and management must be completely delineated, including development of standard instructions, identification of consultants, and identification of appropriate referral centers throughout the state/region.
9) Recommendations and decisions should include consideration of the costs of the screening test, confirmatory testing, accompanying treatment, counseling, and the consequences of false positives. The mechanism of funding those costs should be identified. Expertise in economic factors should be available to those responsible for recommendations and decisions.

6. Estimates of the amount of time that state employees will spend to develop the rule and other necessary resources

The department estimates that it will take approximately 160 hours to develop the proposed rules. This estimate includes time required for research and analysis, coordinating the advisory committee meetings,

rule drafting, preparing any related documents, holding a public hearing, and communicating with affected persons and groups.

7. Description of all of the entities that may be affected by the rule, including any local governmental units, businesses, economic sectors, or public utility ratepayers who may reasonably be anticipated to be affected by the rule

Newborns and parents of newborns will benefit from early diagnosis through newborn screening and follow up treatment.

The state laboratory of hygiene will provide ongoing testing for additional conditions and the WI Newborn Screening Program will have additional newborn screening conditions to follow up and report on.

Expert consultants in the newborn screening field and health care providers will have a slight increase in patient care for those diagnosed through newborn screening.

8. Summary and preliminary comparison of any existing or proposed federal regulation that is intended to address the activities to be regulated by the rule

The department knows of no existing or federal regulation that addresses the activities of this rule.

9. Anticipated economic impact, locally or statewide

The anticipated economic impact is greater than $50,000 but less than $20 million.

The proposed rule may have a moderate economic impact. By adding Pompe disease to the blood card, it increases the cost to the Wisconsin State Lab of Hygiene by $10.49 x 64,000 births amounting to $671,360. The $671,360 would be needed from the state laboratory of hygiene to cover screening and testing. It is estimated that approximately 10 babies will be diagnosed with Pompe Disease per year if the condition was to be added to the screening panel.

10. Agency contacts

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